Hypertrophic cardiomyopathy (HCM) is a genetic cardiovascular disease characterized by a hypertrophied, nondilated left ventricle (LV) in the absence of other causes. HCM is relatively common and is one of the most common causes of sudden death in athletes.1 HCM is genotypically and phenotypically heterogeneous.2,3 The differential diagnosis for HCM includes physiological remodeling seen in athlete’s heart.4 Distinguishing HCM from athlete’s heart is of critical importance. Characteristics favoring HCM and athlete’s heart are listed in the Table.

An 18-year-old man was referred for evaluation after a markedly abnormal ECG (Figure 1A) was discovered at a professional hockey scouting event. He was otherwise healthy and asymptomatic. He had no family history of cardiomyopathies or sudden unexplained death. A contrast enhanced transthoracic echocardiogram showed apical HCM with a maximal LV wall thickness of 24 mm and spade-like appearance (Movie I in the Data Supplement). LV chamber size and systolic function were normal without dynamic obstruction. Diastolic function assessment showed indeterminate diastolic function grade. On cardiac MRI, the apex and anterior septum measured 25 and 13 mm, respectively (Figure 2A; Movie II in the Data Supplement). First pass perfusion was normal, but postcontrast images demonstrated patchy areas of late gadolinium enhancement suggestive of fibrosis (Figure 3A). Exercise ECG and 24-hour Holter monitor showed no ventricular arrhythmias. On genetic testing, a de novo HCM-associated mutation in MYH7-encoded β myosin heavy chain gene at position 530 (p. Ile530Val) was identified. The patient’s parents and 2 siblings, all phenotype negative for genetic testing was repeated to exclude the possibility of a false-positive result, but he was again found to be genotype positive.

He continued to be asymptomatic until 16 months after the diagnosis when he developed presyncope and tachy-palpitations. An event recorder demonstrated episodes of supraventricular tachycardia and nonsustained ventricular tachycardia. Given his symptoms, documented nonsustained ventricular tachycardia, and late gadolinium enhancement on cardiac MRI, an implantable cardioverter defibrillator was implanted. Since the implantable cardioverter defibrillator placement, now >5 years ago, the patient has done well with no appropriate implantable cardioverter defibrillator discharges. On his most recent echocardiogram (6 years and 2 months since diagnosis), maximum thickness remained 12 mm at the apex. These images demonstrate a novel case of LV hypertrophy regression after detraining in a patient with clinical HCM and an HCM-associated mutation. There are limited data supporting regression of LV hypertrophy with pharmacotherapy and surgical relief of outflow tract obstruction, but to our knowledge none on regression after detraining. At initial diagnosis, the extreme ECG changes, marked asymmetrical hypertrophy, late gadolinium enhancement, and genetic mutation were all consistent with an underlying pathological process (ie, sarcomeric HCM) rather than physiological adaptation. However, it is possible the patient has both athletic adaptation to intense training which has currently regressed and HCM.
Alternatively, this case may challenge the existing dogma that regression is only seen in physiological remodeling such as athlete’s heart and that HCM phenotypic conversion is not altered by exogenous loads on the heart. Overall, it is of clinical importance to distinguish HCM and athlete’s heart because of the differing recommendations and prognosis. The potential lifestyle restrictions for HCM have not only physical ramifications but also psychological and financial, as in this patient who was a professional athlete prospect.

Table. Characteristics Favoring HCM and Athlete’s Heart

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<tr>
<th>Favors HCM</th>
<th>Favors Athlete’s Heart</th>
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<tr>
<td>Asymmetrical hypertrophy</td>
<td>Homogenous hypertrophy with maximal thickness of 15 mm</td>
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<td>Diastolic dysfunction</td>
<td>Ellipsoid LV shape</td>
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<td>Late gadolinium enhancement on</td>
<td>Regression with deconditioning</td>
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<td>cardiac MRI</td>
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HCM indicates hypertrophic cardiomyopathy; and LV, left ventricular.

Disclosures

None.

References


Key Words: athletes ◼ cardiomyopathy, hypertrophic ◼ death, sudden ◼ defibrillator, implantable ◼ tachycardia, supraventricular

Figure 1. A, Initial ECG demonstrating left ventricular hypertrophy voltage criteria, ST depression, and deeply inverted T waves consistent with apical hypertrophic cardiomyopathy. B, Follow-up ECG (14 months) with less prominent T wave inversions.

Figure 2. A, Initial cardiac MRI steady-state free precession sequence in a 4-chamber view demonstrating apical hypertrophy (June 2008). B, Follow-up cardiac MRI (August 2009) demonstrating regression of apical hypertrophy.
Figure 3. A. Postcontrast delayed enhancement sequence represented in a long axis view demonstrating the presence of apical hypertrophy and abnormal patchy apical myocardial delayed enhancement (June 2008). B. Follow-up cardiac MRI demonstrating regression of apical hypertrophy but persistence of abnormal myocardial delayed enhancement (August 2009). LGE indicates late gadolinium enhancement.
Hypertrophic Cardiomyopathy, Athlete's Heart, or Both: A Case of Hypertrophic Cardiomyopathy Regression
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SUPPLEMENTAL MATERIAL

Video Legends

Video Clip 1. Initial 2D contrast enhanced echocardiography in a four chamber view demonstrating apical hypertrophy (June 2008).

Video Clip 2. Initial cardiac MRI in a three chamber view demonstrating apical hypertrophy (June 2008).

Video Clip 3. Follow-up cardiac MRI in a three chamber view showing regression of hypertrophy (Aug 2009).

Video Clip 4. Follow-up non-contrast enhanced echocardiography in a four chamber view showing regression of hypertrophy (Aug 2009).